CASE REPORT

Recurrent Dermatofibrosarcoma Protuberans- A Rare Entity, Two Case Reports in a Tertiary Care Center of North India

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ABSTRACT

We present two typical cases of dermatofibrosarcoma protuberans (DFSP) with a complaint of recurrence, make a review of the most important aspects. This is a rare tumor and we call special attention to the fact that its recurrence is extremely frequent, so there is an absolute need to observe these patients periodically after surgery.

Both cases were operated by wide local excision with tumor-free margins and sent for radiotherapy. The histopathological reports confirmed the recurrence of dermatofibrosarcoma protuberans(DFSP). The patients were followed for 3 months but no signs of recurrence seen after excision of recurrent tumor mass

This is a rare tumor and we call special attention to the fact that its recurrence is extremely frequent, so there is absolute need to observe these patients periodically after surgery. These cases motivated us to review the most important aspects of this rare tumor, besides presenting an interesting and typical case of DFSP with recurrence. All dermatologists and surgeons must know about the frequent recurrence of this tumor, sometimes even when excised with wide margins. For this reason, these patients must be observed periodically after surgery for a long time.

Keywords: Recurrent dermatofibrosarcoma protuberans, DFSP, Soft tissue tumours, Recurrence, Case report.

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INTRODUCTION

Dermatofibrosarcoma protuberans (DFSP) is a rare soft tissue tumor with an incidence rate of 4.2–4.5 cases per million persons per year and local recurrence rates up to 60%. It grows slowly and usually presents as a nodular superficial lesion on the trunk or the extremities. Although these tumors are locally aggressive, with a high recurrence rate following surgery, the prognosis is considered excellent when effectively treated. In this case report, we discuss a case of recurrent DFSP of the inguinal region after a latency of 7 years and its surgical management in detail.

CASE SUMMARY 1

A 44-year-old male from Northern India presented to the Surgical Outpatient Department with a gradually increasing nodule of 5 cm in the right inguinal region. It was not associated with any pain, ulceration or discharge. He had a similar complaint in the same location 7 years ago and was diagnosed with biopsy-proven DFSP and was surgically treated for the same. He did not have the records of his previous surgery, but he had the histopathology report, which stated the diagnosis of DFSP with histologically negative margin status. According to the history given by the patient, he did well after the surgery and was disease-free until he noticed this nodule few months ago. There was no history of radiotherapy and/or chemotherapy. He did not have any other co-morbidities. There was no family history of breast, colon or skin cancer.

He had a normal body mass index and his vital parameters and systemic examination was within normal limits. On examination, a 5.5 × 5 cm firm reddish-pink nodule, fixed with the overlying skin and restricted mobility, was present at right inguinal region. Since it was present over his previous scar, the anatomy of the scar was obscured. There was no inguinal lymphadenopathy. Left inguinal region was normal.

His complete blood count, liver function test and kidney function test were within normal limits. A chest radiograph and contrast-enhanced magnetic resonance imaging (MRI) pelvis scan were performed to evaluate local extension and metastasis, which were absent. Fine-needle aspiration cytology of the nodule revealed spindle cells (Figure 1). After a careful history, examination and diagnostic workup, a preoperative diagnosis of recurrent DFSP was made. Keeping the aggressive nature of the recurrent tumour and future reconstruction

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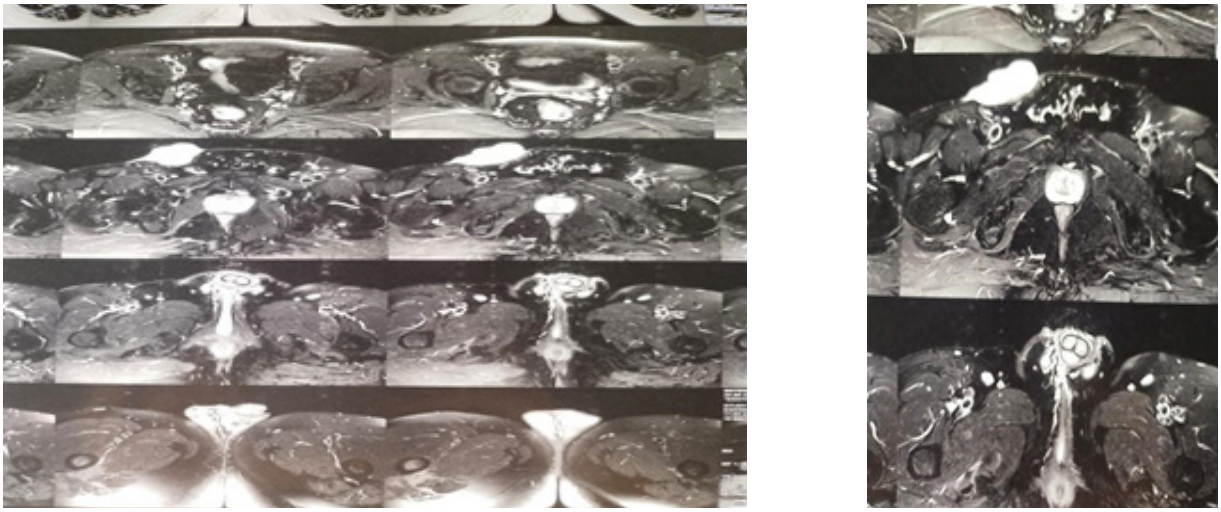

Figure 1: MRI showing well defined multilobulated T2 hyperintense and T1 hypointense superficial mass lesion seen in skin and subcutaneous tissue.

in mind, a wide local excision (WLE) was planned, after consultation with the Plastic Surgery Department.

An elliptical incision was made around the tumor, taking 2.5 cm margin, including the previous surgical scar. A wide local excision of the tumor mass was done. The gap was closed primarily by interrupted mattress sutures.

The wide local specimen measured 7.5 × 5.0× 4.0 cm, showing skin covered raised nodular mass with skin eclipse ms. 7.5 × 5.0 cm. Cut. surface shows whitish smooth, soft mass measuring 4.0 * 3.0 * 2.5 cm. Multiple section from lobulated masses shows the uniform population of slender fibroblast in a distinct monotonous storiform pattern. There is mild nuclear pleomorphism with low mitotic activity. The tumor is infiltrating the dermis and subcutis reaching upto epidermis without uninvolved dermis zones in most areas. The overlying epidermis is unremarkable to atrophic. Foci of subcutaneous fatty tissue and adenexal elements are trapped within the tumor whorls. Inflammatory cells and xanthoma cells are frequently seen suggestive of dermatofibrosarcoma protuberans.

The post-operative period was uneventful with good functional results. The patient was then referred to the Medical Oncology Department for consideration of imatinib therapy and/or radiotherapy. There were no signs of recurrence at 2 months follow-up.

CASE SUMMARY 2

A 22-year-old female from Northern India presented to the Surgical Outpatient Department with a gradually increasing nodule of 4 cm in the right breast region. It was not associated with any pain, ulceration or discharge. She had a similar complaint in the same location 2 years ago and was diagnosed with biopsy-proven DFSP and was surgically treated for the same. She did not have the records of her previous surgery, but she had the histopathology report, which stated the diagnosis of DFSP with histologically negative margin status. According to the history given by the patient, she did well after the surgery and was disease free until she noticed this nodule few months ago. There was no history of radiotherapy and/or chemotherapy. She did not have any other co-morbidities. There was no family history of breast, colon or skin cancer.

She had a normal body mass index and her vital parameters and systemic examination was within normal limits. On examination, a 3× 2-cm firm reddish-pink nodule, fixed with the overlying skin and restricted mobility, was present at right breast region (Figure 1). Since it was present over her previous scar, the anatomy of the scar was obscured. There was no axillary lymphadenopathy. Left axillary region was normal.

Her complete blood count, liver function, and kidney function test were within normal limits. A chest radiograph was performed to evaluate local extension and metastasis, which were absent. Fine-needle aspiration cytology of the nodule revealed spindle cells. After a careful history, examination and diagnostic workup, a preoperative diagnosis of recurrent DFSP was made. Keeping the aggressive nature of the recurrent tumour and future reconstruction in mind, a wide local excision (WLE) was planned, after consultation with the Plastic Surgery Department (Figure 2).

An elliptical incision was made around the tumour, taking 2.5 cm margin, including the previous surgical scar. A wide local excision of tumor mass was done. The gap was closed primarily by interrupted mattress sutures (Figure 3).

The post-operative period was uneventful with good functional results. The patient was then referred to the Medical Oncology Department for consideration of imatinib therapy and/or radiotherapy. There were no signs of recurrence at 2 months follow-up (Figure 4).

DISCUSSION

DFSP is an uncommon, low-grade sarcoma of dermal fibroblast origin with an incidence rate of 4.2–4.5 cases per million persons per year. The median age of presentation is 38.5 years with roughly equal distribution between males and females. The trunk and extremities are considered the most common sites for DFSP; nevertheless, it has been described in various

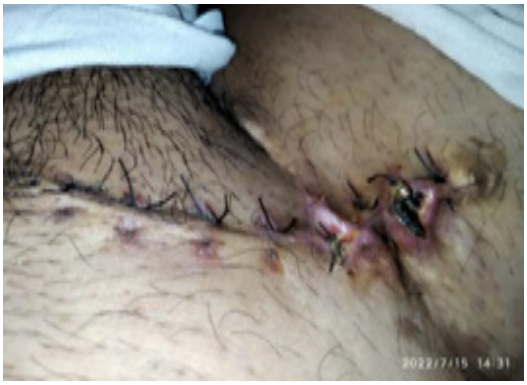

Figure 2: Post WLE.

parts of the body, including the neck, head, breast and even vulva.

DFSP has been reported to arise in areas with a history of prior trauma, including tattoos, vaccination sites, burn scars, surgical scars and radiation treatment. The exact mechanism in which trauma may predispose for the development of DFSP is unknown, but it seems intuitive that chronic inflammation and stimulation of the immune system at a local level may trigger the immunopathologic changes that could lead to the malignant transformation of dermal cells. Studies have shown that DFSP demonstrates the chromosomal translocation *t*(17; 22) (*q*22; *q*13) between chromosomes 17 and 22, which is thought to be a key in the tumor's pathogenesis. The chromosomal translocation leads to the fusion of the platelet-derived growth factor (PDGF) beta polypeptide gene and collagen type 1A1 gene, resulting in the overproduction of PDGF and eventually cellular proliferation and tumor formation. This chromosomal translocation is present in over 90% of DFSP and can be detected either by fluorescence *in-situ* hybridization (FISH) on interphase nuclei and/or by multiplex reverse transcription-polymerase chain reaction (RT-PCR). Other chromosomal translocations like the COL6A3-PDGFD fusion gene have been associated with an apparent predilection for breast. This particular translocation is not detected by conventional RT-PCR used for DFSP and requires RNA sequencing and FISH for detection.

The early clinical symptoms of DFSP are non-specific, making diagnosis difficult and leading to a high incidence of misdiagnosis. The most frequent presentation described in adults is a large plaque presenting multiple nodules on its surface. Due to lack of pathognomonic clinical findings, DFSP can be mistaken for a keloid, hypertrophic scar, sebaceous cyst or lipoma and is often referred late for specialised evaluation. In our case, the lack of characteristic clinical appearance of keloid/hypertrophic scar as well as prior history of DFSP with irregular growth raised suspicion of recurrence. Therefore, a history of prior trauma associated with an ill-defined cutaneous lesion warrants consideration of DFSP in the differential diagnosis.

Pathological and immunohistochemical examinations are currently the gold standard for diagnosing DFSP. Microscopically, it demonstrates a uniform population of

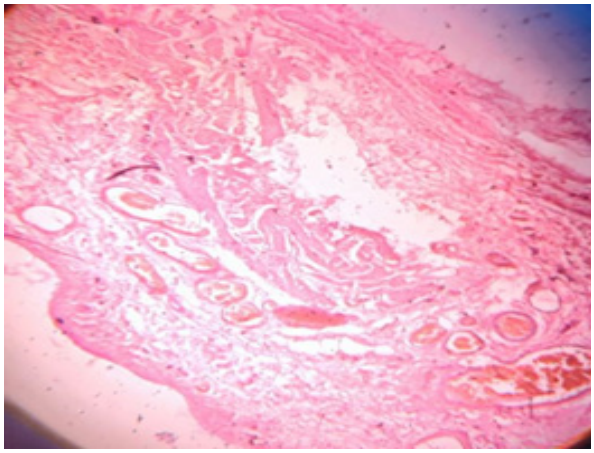

Figure 3: Uniform population of slender fibroblast in a distinct monotonous storiform pattern ; foci of subcutaneous fatty tissue and adnexal elements are trapped within the tumour.

monomorphic spindle cells arranged in a characteristic storiform pattern over a background of fibrous stroma. These cells may show infiltration into the surrounding subcutaneous fat. The lesions show little nuclear pleomorphism and have low to moderate mitotic activity. They occasionally show myxoid or densely collagenous areas or areas of hemorrhage. Three-dimensional reconstruction of DFSP has shown tumours with highly irregular shapes and frequent finger-like extensions. As a result, incomplete removal and subsequent recurrence are common. The local recurrence rate for DFSP is reported up to 60%. Several histopathological variants of DFSP have been described including pigmented DFSP or Bednar tumour, myxoid, juvenile DFSP or giant cell fibroblastoma, atrophic, sclerosing and myoid, occurring in pure form or admixed with one of the others creating hybrid lesions. Fibrosarcomatous DFSP is also an atypical DFSP subtype, with high rates of recurrence and metastasis. Immunohistochemical studies reveal strong staining with human progenitor cell antigen CD34, but negative for factor XIIIa.

The role of radiological imaging is limited in the management of DFSP. Most patients undergo excision and biopsy of the mass without the need for CT or magnetic resonance imaging (MRI) due to its superficial location. On CT, the tumours appear as superficial well-defined nodular masses involving the skin and subcutaneous fatty tissue with attenuation values approaching that of skeletal muscle and a moderate degree of contrast enhancement. On MRI, typical lesions demonstrate prolonged T1 and T2 relaxation times (hyperintense on T2 and hypointense on T1 weighted images) and signal characteristics, which are considered non-specific and demonstrated by many other benign and malignant soft tissue condition. Cases of DFSP with atypical locations do not show much radiological difference from that found in the typical common locations such as trunk and extremities. However, imaging assumes an important role in atypical cases or deep-seated lesions for the definition of margins and local invasion as a part of the preoperative analysis. In addition, metastasis, although rare, can be ruled out with imaging. In

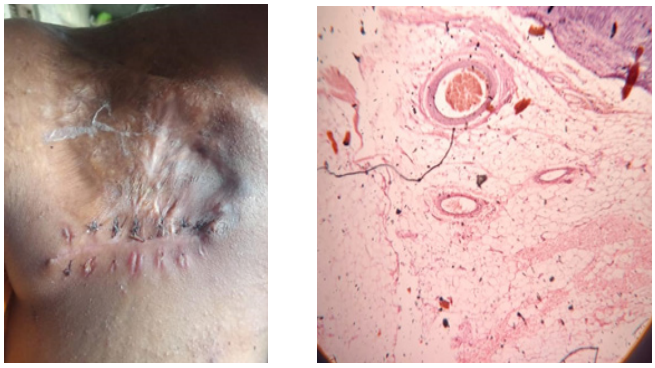

Figure 4: post operative image of rt. mammary region; Another image showing the HPE.

our case, contrast MRI pelvis was performed to assess invasion into surrounding structures as well as to rule out metastasis, as this was an atypical case with recurrence.

The treatment of DFSP is primarily surgical. The size and location of the tumor and cosmetic issues will dictate the most appropriate surgical procedure. Modified Mohs surgery and traditional WLE, typically with 2–4 cm margins to investing fascia that are subsequently verified to be clear through the traditional pathologic examination, are all methods to achieve complete histologic assessment. Moh's surgery offers maximal tissue conservation and better margins as compared to WLE but the local recurrence rates are statistically similar. Mohs micrographic surgery is preferred in regions such as the head and neck, where tissue conservation is important. However, the lack of expertise in all centers results in using the technique of WLE with or without plastic reconstruction as the surgery of choice.

Margin status is important in the surgical management of DFSP as it determines further treatment and prognosis. Disease-free survival after treatment for DFSP is strongly predicted by tumor depth in the primary setting and margin status in recurrent tumors. However, because of its proclivity for irregular and frequently deep sub-clinical extensions, these 'tricky' margins pose a challenge to surgical dexterity. In the present case, as it was a recurrent tumor, we opted for a WLE approach to give us as much margin as possible.

If not given previously, radiation therapy or imatinib mesylate should be considered if this is not possible, or if additional resection would lead to unacceptable functional or cosmetic outcomes. Imatinib mesylate, a protein tyrosine kinase inhibitor, has been approved by the FDA for the treatment of unresectable, recurrent and/or metastatic DFSP in adult patients as it inhibits the overactivity of PDGF receptors in these tumor cells. Because of the recurrent nature of DFSP, our patient was referred for consideration of chemotherapy with imatinib and radiotherapy as additional resection will lead to a functional loss, in case of recurrence in the future.

In a prospective study of 244 patients, the median time to recurrence, local and distal, was 35 months. However, recurrences after long latency have also been reported, including 7 years in the present case. This emphasizes the importance of follow-up in DFSP cases and advocates for a long duration of self-surveillance of surgical scars by patients themselves, as it is not always practical to follow every patient.

REFERENCES

1. NCCN Clinical Practice Guidelines in Oncology for Dermatofibrosarcoma Protuberans. [https://www.nccn.org/professionals/physician_gls/default.aspx]
2. Al Barwani AS, Taif S, Al Mazrouai RA, et al. Dermatofibrosarcoma protuberans: insights into a rare soft tissue tumor. J Clin Imaging Sci. 2016;6:16. doi: 10.4103/2156-7514.181492. - DOI - PMC - PubMed
3. Boukovalas S, Castillo AC, Andry D, et al. Dermatofibrosarcoma protuberans: trauma and genetics. Ann Plast Reconstr Surg. 2017;1(1):1001.
4. Brooks J, Ramsey ML. StatPearls. Treasure Island, FL: StatPearls Publishing; 2018. [2018 Jul 15]. Cancer, dermatofibrosarcoma protuberans. [Internet][https://www.ncbi.nlm.nih.gov/books/NBK513305/]
5. Dickson BC, Hornick JL, Fletcher CDM, et al. Dermatofibrosarcoma protuberans with a novel COL6A3-PDGFD fusion gene and apparent predilection for breast. Genes Chromosomes Cancer. 2018 doi: 10.1002/gcc.22663. - DOI - PMC - PubMed
6. Lyu A, Lyu A. Dermatofibrosarcoma protuberans: a clinical analysis. Oncol Lett. 2018;16:1855–1862. doi: 10.3892/ol.2018.8802. - DOI - PMC - PubMed
7. Stivala A, Lombardo GAG, Pompili G, et al. Dermatofibrosarcoma protuberans: our experience of 59 cases. Oncol Lett. 2012;4(5):1047–1055. doi: 10.3892/ol.2012.887. - DOI - PMC - PubMed
8. Socoliuc C, Zurac S, Andrei R, et al. Multiple histological subtypes of dermatofibrosarcoma protuberans occurring in the same tumor. Rom J Intern Med. 2015;53(1):81–90. doi: 10.1515/rjim-2015-0011. - DOI - PubMed
9. Prabhu VV, Shivani A, Kulkarni SH, et al. Dermatofibrosarcoma protuberans: male breast. Med J DY Patil Univ. 2014;7:85–87. doi: 10.4103/0975-2870.122796. - DOI
10. Swan MC, Banwell PE, Hollowood K, et al. Late recurrence of dermatofibrosarcoma protuberans in the female breast: a case report. Br Assoc Plast Surg. 2005;58:84–87. doi: 10.1016/j.bjps.2004.06.017. - DOI - PubMed
11. Fields RC, Hameed M, Qin LX, et al. Dermatofibrosarcoma protuberans (DFSP): predictors of recurrence and the use of systemic therapy. Ann Surg Oncol. 2011;18(2):328–336. doi: 10.1245/s10434-010-1316-5. - DOI - PMC - PubMed